UCRL-JC-123178 Abs

A NEW ASSAY FOR THE SIMULTANEOUS DETECTION OF ANEUPLOIDIES FOR CHROMOSOMES 21, X AND Y IN HUMAN SPERM BY MULTI-COLOR FISH. A. Baumgartner[1,2], P. Van Hummelen[1], X. Lowe[1], I.-D. Adler[2]* and A.J. Wyrobek[1]. [1]Bio. Biotech. Res. Prog., Lawrence Livermore Natl. Lab., Livermore, CA; [2]GSF-Institut für Säugetiergenetik, Neuherberg, Germany.

The majority of aneuploidies detected among liveborn offspring involves chromosomes 21, X or Y. We developed a multi-color FISH method for simultaneously detecting sperm carrying numerical abnormalities for these three chromosomes. This method was evaluated using selected semen samples previously analyzed with a separate X-Y-8 FISH aneuploidy assay and with the hamster-egg technique for sperm cytogenetics. A preliminary evaluation of this assay was conducted to determine the frequency of disomy and diploidy in semen of healthy donors. The sex ratios were not significantly different from 1:1 among 22,916 sperm from two donors. The frequencies of sperm disomic for chromosome 21 were 2.4 and 8.9 per 10^4 nuclei, which was not statistically different from the frequency of chromosome 8 disomy detected by the X-Y-8 FISH assay. The frequencies of sperm disomic for the sex chromosomes (XX, YY and XY) were 16.5 and 15.7 per 10^4 nuclei while the diploidy frequencies were 10.2 and 16.7. The disomy frequency for chromosome 21 determined with the X-Y-21 assay was similar to that determined in previous studies using the hamster-egg technique: 12 per 22,916 (FISH) vs. 5 per 5,997 (hamster technique) with $p=0.378$. These findings provide initial validation for the aneuploidy frequencies obtained with the new X-Y-21 sperm FISH assay. Future studies will investigate aspects of donor variations and effects of exposure to toxicants.

[Work was performed under the auspices of the US DOE by the Lawrence Livermore Natl. Lab. under contract W-7405-ENG-48; A.B. was supported by EU Contract EV5V-CT94-0403]